HOT SPOT

Hang on Tight—Stories, Parables, Occurrences, Training

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MORTALITY ALERT!

COULD THIS HAPPEN TO YOU?

A very outgoing and likable individual, in their mid forties is found dead. Death occurred some time in the early morning hours. There was a history of early difficulty with weight gain with subsequent obesity. This individual was short in stature, only 59 inches tall, and weighed 265 pounds. The hands and feet were small and other features consistent with the Prader-Willi syndrome were present.

Prader-Willi Syndrome (PWS) is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity. Most cases of PWS are due to a lack of several genes on one of an individual's two chromosome 15s. Suspicion of the diagnosis is first assessed clinically, then confirmed by specialized genetic testing. Genetic testing is important to prevent unnecessary evaluations, assure appropriate management, and allow genetic counseling. Although considered a "rare" disorder, Prader-Willi Syndrome is one of the most common conditions seen in genetics clinic and is the most common genetic cause of obesity that has been identified.

Characteristics typically seen in an adult with Prader-Willi Syndrome may include:

- Decreased vomiting
- Esotropia (cross-eyed), myopia (nearsightedness)
- Facial features: narrow bifrontal diameter, almond-shaped eyes, down-turned mouth, small appearing mouth with thin upper lip
- High pain threshold
- Hyperphagia (excessive appetite) due to a hypothalamic abnormality
- Hypogonadism (incomplete pubertal development, infertility is the rule)
- Hypopigmentation (fair hair, eyes, and skin as compared to rest of the family)
- Narrow hands with straight ulnar border
- Osteoporosis
- Scoliosis/kyphosis
- Short stature
- Skin picking
- Sleep disturbance/sleep apnea
- Small hands and feet for height/age
- Speech articulation defects (slowed/slurred/nasal due to low muscle tone)
- Thick, viscous salvia with crusting at corners of the mouth
- Unusual skill with jigsaw puzzles

Individuals with Prader-Willi Syndrome tend to have specific behavior problems. These may include:

- Obsessive/compulsive behaviors (such as collecting and hoarding of possessions, picking at skin irritations, and a strong need for routine and predictability).
- Manic-depressive illness
- Argumentativeness
- Perseveration (repetitive thoughts and verbalizations)
- Moodiness
- Impulsivity
- Compulsivity
- Non-compliance
- Rigidity/ Stubbornness (inflexible, incapable of changing or being changed)
- Tantrums
- Issues of food intake and weight control
- Stealing (usually food or money to buy food)
- Lying
- Pica (eating of inedible objects)

Alerts for individuals with Prader-Willi Syndrome may include:

- Abdominal distention or bloating, pain, and vomiting may be signs of life-threatening gastric emergency
- Adverse reactions to some medications (prolonged and exaggerated responses to standard dosages of medications)
- Dental problems due to poor oral hygiene, thick sticky saliva, soft tooth enamel, teeth grinding, or infrequently rumination

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- Diabetes mellitus, type II may occur secondary to obesity
- Fever may be absent despite serious infection
- Increased risk for respiratory difficulties (due to weak chest muscles)
- Infection of open sores due to skin picking
- Lack of typical pain signals may mask the presence of infection or injury
- Obesity results if total caloric requirement is not decreased due to low metabolic rate
- Other obesity related problems include hypoventilation, hypertension, stasis ulcers, cellulitis, and skin problems in fat folds
- Subtle changes in condition or behavior should be investigated for medical cause
- Supervision required at all times in all settings where food is accessible
- Tendency to bruise easily
- The presence of vomiting may signal a life-threatening illness
- Water intoxication from excess fluid intake or use of certain medications

To date, no medication or surgical intervention has been found helpful in most cases for long-term weight control. A nutritionally sound, restricted calorie diet and an exercise program are critical to weight control. The environment must be designed so there is limited access to food. This may include a lock on the kitchen or the cabinets and refrigerator. Individuals with PWS have a flaw in the hypothalamus part of the brain, which normally indicates a feeling of hunger and feeling full. They have a continuous urge to eat that can not be controlled. Adults with Prader-Willi Syndrome can control their weight best in a home designed specifically for individuals with this syndrome, where food access can be restricted without interfering with the rights of those who do not need such restriction. Without such control, 95 percent of individuals with Prader-Willi Syndrome develop weight problems. Failure to restrict access to food is equal to medical neglect. Access to food is a strong desire for individuals with PWS. If not restricted to food access, it may become a dangerous and medically neglectful practice, leading to medical emergencies or to premature death related to complications of obesity. Restricting food is the standard of care for an individual with Prader-Willi Syndrome. Until there are interventions for this eating behavior, structured environments with restricted access to food must be standard care for those with Prader-Willi Syndrome.

Why have genetic studies?

A syndrome is defined as the group or recognizable pattern of symptoms or abnormalities that indicate a particular trait of disease. Many syndromes have a genetic basis. Genetic testing may help to show "why" an individual has a tendency to get a certain disorder. Once identified, a course for medical care can be determined. Gene testing may help scientists determine if gene mutations work alone, or in combination with other mutations or with environmental factors to cause disease. Genetic testing gives a hope that cures may ultimately be possible. It is hoped that discovery of a gene causing a disease in certain families may give scientists the lead they have been searching for to reach a cure. Genetic counseling is a service to help individuals and families translate scientific knowledge into practical information. Genetic counseling helps to identify and understand what particular traits may be passed on to children and also to determine particular risks that may influence pregnancy outcomes. Counselors or the physician can refer to a resource in the community that deals with a specific genetic condition, or to a medical specialist, educational specialists or family support groups.

Gene tests (also called DNA-based tests) are procedures used to test for genetic disorders. Testing involves direct inspection of the DNA molecule. Genetic tests are used for several reasons, including:

- Carrier screening (identifying individuals who carry one copy of a gene for a disease that requires two copies for the disease to be expressed)
- Prenatal diagnostic testing
- Newborn screening
- Testing for predicting adult-onset disorders (e.g. Huntington's disease)
- Testing for estimating the risk of developing adult-onset cancers and Alzheimer's disease
- Confirm a diagnosis for a symptomatic individual
- Forensic/identity testing

One reason why families get a genetic study is to learn if a specific disability may return within a family. The family can be counseled in relation to their own medical and family history. Genetic testing can also offer typical profiles for learning strengths and weaknesses. In this way, there may be a helpful framework for assessing individuals specific needs. Acknowledging the existence of syndrome-specific behaviors can provide new insights into treatment. Having a specific diagnosis may provide a starting point. Families need to know that many conditions are genetic (they involve a person's genes) and may not necessarily run in families.

The comprehensive genetic evaluation for an individual with developmental disabilities involves a careful examination of many things. It may include the physical features of the individual, as well as a review of the prenatal, medical, developmental, and family history. Diagnostic, laboratory and other studies are also possible. All of these evaluations help to reveal the patterns upon which a syndrome diagnosis is based. A syndrome is defined as the group or recognizable pattern of symptoms or abnormalities that indicate a particular trait or disease. For individuals who never had, or underwent genetic evaluations more than five years ago, it may be desirable to be re-evaluated to keep up with changing technology. For many diseases, genetic information will help the individual and the physician weigh the risks and benefits of different treatments.

Gene testing has improved lives. Some tests are used to clarify a diagnosis and direct a physician toward treatments. Other tests may identify individuals at high risk for conditions that may be preventable. Some tests give only a probability for developing a specific disorder. Interpreting results of any testing is difficult because some people who carry a disease-associated gene may never develop the disease.

Clinical genetics centers exist in every state. They are often affiliated with major university hospitals. There are a number of different

genetic services available. The local chapter of the March of Dimes has information on services in the local area as well as the physician or the nearest hospital associated with a medical school.

Genetic counseling describes a process where families concerned about birth defects or genetic diseases get support to make informed reproductive decisions. This process involves a genetic counselor (a health care professional specialized in medical genetics and counseling), and a geneticist (a research scientist or physician who makes medical and laboratory diagnoses).

Clinical genetics evaluation is the medical work-up of a family with a potential or known genetic condition. A clinical geneticist is a physician with knowledge in recognizing the symptoms and causes of genetic disorders and birth defects.

Diagnostic genetics laboratories exist at many hospital-based centers and provide genetic counseling services. A cytogenetics laboratory performs chromosome studies. A molecular (DNA) lab tests for a single gene disorder.

Specialty clinics (based most often at university medical centers and children's hospitals), offer specialists for a particular disorder using a team approach to best meet the medical and therapeutic needs of an individual (e.g. clinics for PKU, Down syndrome, fragile X, or spina bifida).

Genetic support groups exist to provide emotional support for families with genetic conditions.

METHODS FOR INFECTION CONTROL

A routine schedule for cleaning should be established and carried out by a designated person. Places for food preparation, bathroom facilities, and any other special areas identified must be cleaned and disinfected on a daily basis. A regular cleaning schedule for other areas, such as classrooms, workshops, and recreational settings, should be established. Cleaning products that will kill or inactivate both bacteria and viruses should be used. The Environmental Protection Agency (EPA) publishes a list of approved disinfectants that is updated periodically. It can be obtained by contacting the local health department. The local health department can provide guidelines and information on regulations concerning the preparation, storage, and serving of foods.

WINTER STORMS

A major winter storm can be lethal. Preparing for cold weather conditions and responding to them effectively can reduce the dangers caused by winter storms. Listen to local forecasts to determine any impact the weather may have on your schedule. The National Weather Service may issue the following advisories.

- **Winter Storm Watch** indicates that severe winter weather may affect the local area. Alerts the public to the possibility of a blizzard, heavy snow, freezing rain, or heavy sleet. Watches are usually issued 12 to 36 hours before the beginning of a Winter Storm. During a snowstorm watch, stay tuned for developing weather conditions. Travel only if necessary. Make sure there is proper ventilation if using a portable heater.
- **Winter Storm Warning** indicates that severe winter weather conditions are definitely on the way. Warnings are usually issued 6 to 24 hours before the event is expected to begin. A **Blizzard Warning** means that large amounts of falling or blowing snow and sustained winds of at least 35 miles per hour are expected for several hours. During a snowstorm warning, stay indoors until the WARNING is lifted. If you have to go outside, be sure to dress in several layers of clothes. Be careful walking on snowy, icy sidewalks.
- **Traveler's Advisory** indicates that severe winter conditions might make driving difficult or dangerous. Avoid traveling in a storm if at all possible. If you must travel, remember the following:
  - Keep gas tank full for emergency use and to keep the fuel line from freezing.
  - Let someone know where you are going and when you should get there.
  - Have emergency supplies in the car, including a flashlight, flares, and blankets.
- **Wind Chill Advisory** is issued when wind chill temperatures are expected to be between 20 below and 34 degrees below zero. Wind Chill is the term used to describe the rate of heat loss on the human body resulting from the combined effect of low temperature and wind. As winds increase, heat is carried away from the body at a faster rate, driving down both the skin temperature and eventually the internal body temperature. Exposure to low wind chills can be life threatening to both humans and animals. Stay indoors if possible, if you must go outside; be sure to dress appropriately.

DYING WITH DIGNITY. COULD THIS HAPPEN TO YOU?

Not long ago an 82-year-old individual experienced several extreme medical complications including a stroke, congestive heart failure, irregular heart beat, recurrent pneumonia and fluid retention. During this hospitalization, a meeting was held to discuss the "code" status for this person. This simply meant that there was interest in having a discussion of a "Do Not Resuscitate" (DNR) status as opposed to a "full code" status. Currently there is a policy available which discusses this issue in the community and developmental centers (DMRS 2000-71 (REVISED) Life Sustaining Treatment). It appears that during this time period the Independent Support Coordinator and the conservator were working out the legality of having a DNR status for this individual. Several months after this initial meeting the doctor requested a meeting with the legally responsible party to discuss the status of this individual. One week later the doctor again requested a time to discuss the DNR status. One month after this request, another plea from the doctor to discuss the status. Five days later this individual died after undergoing a "full code" status to include CPR, cardiac medications and intubation. Recognizing the right of every person to die a natural death and providing dignity to a dying person is consistent with humane care and treatment, and with the highest standards of accepted medical practice. Choices concerning a Do Not Resuscitate status for an individual are a difficult yet necessary part of the decision making process for the person who is legally responsible for that individual.

TRAINING AVAILABLE AS OF JULY 2001

1. Advance Care Planning
2. Breast Examination
3. Comprehensive Health Care Assessments
4. Health Care Plans
5. Health Passport
6. Health Risk Screening/Physical Status Review/PSR
7. How to Ask Questions to Improve Health Care
8. Medication Administration for Unlicensed Personnel
9. Medication Administration for Unlicensed Personnel Recertification
10. Medication Error and Omissions Form
11. Orientation to Community Developmental Nursing
12. Psychotropic Medication Administration Policy Training
13. Psychotropic Medication Training (drug information sheets)
14. Seizure Training
15. Supporting People with Developmental Disabilities in the Health Care System
16. When to Seek Medical Attention

TRAINING AVAILABLE AS OF JULY 2001 OFFERED ON AN INDIVIDUAL BASIS

1. Diabetes Mellitus
2. Diabetes Mellitus and Insulin Administration
3. Epi-Pen
4. Medication Self Administration

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Ideas for future issues are greatly appreciated.

Please feel free to submit any news articles or request any information on issues that will promote a better understanding of the MR/DD population. Contact Ruth Givens at 615-532-6547 or by E-mail at rgivens@mail.state.tn.us

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